ASC Guide for Questions from Providers on NBS CF Results of:

Elevated IRT and Negative CFTR DNA Panel

- The interpretation of these results is **Screen Negative for Cystic Fibrosis;** no further action or follow-up is required.

- An elevated IRT (Immunoreactive Trypsinogen) is **not** diagnostic for CF. The majority of elevated IRT results are false positives.

- Trypsinogen, a precursor to the enzyme trypsin, is produced in the pancreas. Blockage of pancreatic ducts causes elevations of trypsinogen in the blood of most newborns with CF. Newborns who do **not** have CF can have temporary elevations of trypsinogen; their levels become normal within a few weeks.

- Elevated IRT levels may be due to the biological and genetic variability of newborns, and some studies have found high IRT levels associated with asphyxia during birth (measured by Apgar score), chronic fetal distress, infection or other illnesses requiring neonatal intensive care. How these factors influence IRT levels is not completely understood.

- The chance that an infant with an elevated IRT and negative CFTR DNA panel actually has CF is low.

- If the baby has symptoms of CF, both parents are known carriers of CF or if there is a family history of CF, consultation with a CF Center specialist about diagnostic testing and evaluation for the infant is recommended.

  Symptoms of CF in the first few months of life may include some of the following:
  - ◊ meconium ileus
  - ◊ very salty sweat
  - ◊ failure to thrive, slow growth
  - ◊ recurrent respiratory infections, including respiratory syncytial virus (RSV) infections
  - ◊ malnutrition (vitamin deficiencies, malabsorption)
  - ◊ frequent runny stools

- For all the disorders screened for, the newborn screening result should not be considered diagnostic, and cannot replace the individualized evaluation and diagnosis of an infant by a well-trained, knowledgeable health care provider. A negative screening result does not rule out the possibility of a disorder. Health care providers should remain watchful for any sign or symptoms of the disorders screened for.